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# *Editorial Comment*

# More on the Noonan-CFC Controversy

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We read with interest the report by Lorenzetti and Fryn [1996] on a Noonan syndrome patient with retinitis pigmentosa, published in this issue of the Journal. We definitely concur with their diagnosis. Actually, the patient has all of those characteristics that we consider distinctive of Noonan syndrome vis-à-vis CFC syndrome, i.e., thick hair (not as sparse and woolly as in CFC syndrome), bushy eyebrows (not sparse or absent, as in CFC syndrome), absence of skin lesions (only dryness is mentioned by the authors), neck webbing (rarely reported in CFC syndrome), and normal intelligence.

Therefore, the authors' claim, that this patient supports the view that Noonan and CFC syndromes may be the same entity, only rests on the finding of ocular abnormalities. However, those abnormalities listed in their Table I are admittedly common to both syndromes and are at least partially attributable to congenital lymphedema and muscular hypotonia, which are likely to represent common pathogenetic pathways in the phenogenesis of Noonan and CFC syndromes [Mendez, 1985; Neri et al., 1991]. This leaves retinitis pigmentosa as the sole potentially critical and discriminating finding. However, it must be noted that this abnormality has only been reported in the patient of Lorenzetti and Fryns [1996]. In patients described by Dunya et al.[1993] and Adès et al. [1992], the situation seems to be quite different. A "cone dystrophy" is reported in the former, and "thinning of the retinal pigment" in the latter.

This is not nearly enough evidence to conclude that Noonan and CFC are one and the same condition. Even less does the patient of Lorenzetti and Fryns support the contention of Fryer et al. [1991] that CFC and Noonan are examples of contiguous gene syndromes. Again, the issue will be resolved once and for all by the cloning of Noonan and/or CFC gene(s), an accomplishment that hopefully is not too far away, given the recent assignment of a Noonan syndrome locus to 12q [Jamieson et al., 1994].

Meanwhile, we agree with Lorenzetti and Fryns that "ophthalmological investigations are to be recommended in patients with CFC and NS syndrome," including fundoscopy and electroretinography.

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